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ABSTRACT

More and more people are touched daily by genetic information. Even with the technological accomplishments generated by the Human Genome Initiative, genetic information will continue to produce uncertainty. It is this uncertainty that gives rise to many of the complex clinical dilemmas facing genetic and psychology professionals today. Some of the issues facing genetic research today include: (1) how genetic information influences the development of personality and identity formation of people with genetic diseases compared to individuals at risk; and (2) how knowledge about genetic information and the potential of having a life-threatening disease influences family relationships and life expectations. It suggests that overall, psychologists will need to better attend to the clinical presentation of genetic issues with clients. Given the logistics and training limitations of genetic counselors, psychologists must be prepared to work with individuals, couples, and families to explore how the impact of genetic information influences coping, adjustment, and understanding of oneself. They must be open to exploring the intrapsychic issues related to uncertainty, risk perception, and decision-making. (Contains 20 references.) (JDM)

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Genetics and Psychology at a Crossroad: A Road Less Traveled?

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Contrary to popular belief, genetic conditions are relatively common. Medical progress in the treatment and prevention of other diseases has meant today in the U.S. America, 3-5% of all pregnancies result in a child born with birth defects. Genetic conditions account for approximately 25% of reported infant deaths, 40% of childhood mortality, and 5-10% of all pediatric hospital admissions. A genetic basis for all hospital admissions ranges from 25-60%. Many chronic diseases, including diabetes, cancer, hypertension, schizophrenia, and some forms of depression have identified genetic contributants (Kelly, 1986). Given the advances in genetic diagnostic technologies, the possibilities for genetic screening and treatment have greatly expanded.

The increasing visibility of genetics within mental and medical health care has been due to the advent of expanding genetic knowledge and technical expertise derived from efforts of the Human Genome Initiative, increasing awareness of genetic diseases, and disproportionate resource allocations for genetic disease treatments due to their chronic nature. Growing knowledge of the genetic nature of diseases has also matured into a recognized specialty of medical practice. From modest beginnings of "genetic hygiene," the field of medical genetics and genetic counseling today have been elevated to departmental status in many tertiary and secondary medical centers.

The advancements in genetic technology and medical interventions have also created new challenges for individuals, families, and society as a whole. For example, pregnancies once assumed normal until proven otherwise are now viewed as high risk for birth defects until ruled out by genetic tests. Due to the availability of genetic technology, a child with a birth defect once considered an unfortunate stroke of fate is now the responsibility of parents. The possibility to predict the risks of developing serious diseases (e.g. breast cancer, diabetes) through genetic technology has also raised ethical dilemmas and unforeseen psychological consequences for those who wish to obtain their genetic information.

Genetic diseases are not homogeneous. Variability exists in gene expression, age of onset, physical and/or mental sequelae. Some genetic conditions are life-threatening, disfiguring, cause progressive degeneration, and difficult to manage; others are benign, innocuous, static, and have unnoticeable daily consequences. Overall, genetic diseases fall under three broad categories according to the age of onset symptoms appear. Genetic conditions manifesting prenatally are essentially untreatable and often abort spontaneously (e.g, Trisomy 18). Perinatal or early childhood presentation of genetic diseases often respond to interventions if diagnosed promptly and treatment compliance is successful. For example, inborn errors of metabolism may be undetected *in utero* because the dysfunctional metabolism is physiologically compensated by the mother during fetal life. The pathology of the genetic disease manifests only after the infant is separated from its

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mother and must rely on its own defective metabolic resources. Genetic diseases expressed during puberty (e.g., hypertension, diabetes) can be genetically-based or have a genetic component. Most of these diseases are multifactorial with genetic factors and environmental influences contributing to gene expression. Such multifactorial disorders are by far the most common diseases in which genetics plays a role. The fact their development interacts with environmental factors makes them in principle - and to a considerable degree, in practice - more open and responsive to therapeutic interventions although cures are never possible.

> Disclosure of genetic information often involves sharing unpleasant, even devastating information to individuals. Clients often learn of poor prognoses, limited if any treatment options, and diminished life expectancy of loved ones. As technology continues to expand and govern genetic knowledge, so does the influence of geneticists, genetic counselors, and psychologists in the lives of people who seek counseling about health issues about themselves, their children, and/or other family members.

Genetic Counseling and the Psychological Meanings of Genetic Disease

For most people, genetic conditions hold particularly negative connotations. People often perceive genetic diseases as irreversible, contagious, chronic, family-linked, and stigmatizing. Genetic conditions also evoke strong emotions of people such as fear, pity, guilt, and anger. The strong ties between cultural and health-related schemas also burden genetic conditions with cultural connotations. For example, depending on the cultural context, parents of children with genetic defects may perceive their child's problems as punishment for their deeds, and for others a test of their faith. Thus, genetic counselors and psychologists must be knowledgeable and aware of the cultural context of themselves and their clients.

> Genetic counseling is a psychoeducational process which addresses issues regarding risks of having children with birth defects and/or mental retardation (American Society of Human Genetics, 1975; Kelly, 1986; Leroy, 1993). Within this context, clinicians who practice genetic counseling focus on problems and issues associated with genetic diseases in families. Geneticists impart information about medical diagnostics and management, explore decision-making based on risk perception, examine possible options for coping with recurrence of birth defects, and help individuals and their families make psychological and/or physical adjustments to disabilities (American Society of Human Genetics, 1975; Kelly, 1986; Marks, Heimler, Reich, Wexler, & Ince, 1989). Ideally, genetic counseling focuses on a client's learning, understanding, decision-making, bereavement, and coping issues associated with the occurrence or at risk status for genetic disease.

Genetic counseling essentially involves issues of uncertainty. One consequence of this uncertainty is psychological distress. As Leventhal, Diefenbach, and Leventhal showed (1992), interactions between affect, illness, cognitions, and behaviors can be enormously affected. For example, as a means for coping, genetic at-risk individuals were reported to have disturbing and dysfunctional behaviors (e.g, constant self-surveillance of disease symptoms of themselves and other family members), distress (e.g, extreme anxiety at every benign sign), and survivor guilt (e.g, feelings of ambivalence and culpability when

a family member is diagnosed with a genetic disease).

Jemmott, Croyle, & Ditto (1988) have also discussed how people minimize the significance of threatening information through cognitive information processing and cognitive biases. For example, Markova, Forbes, Aledorf, Inwood, Mandalaki, Miller, & Pittadaki (1986) and others (Ekwo, Kim, & Gosselink, 1987; Sagi, Shiloh, & Cohen, 1992; Taylor & Loebel, 1989) showed individuals tended to compare their own situation to more severe cases as a means to minimize health-related information. Sagi et al. (1992) examined the influence of perception and severity of cleft lip and/or palate (cleft lip and/or palate is a multifactorial congenital malformation in which the palate and lip fail to close). They showed that parents of children with a cleft lip and/or palate perceived their child's own birth defect as less severe than they viewed the same birth defect in general.

Risk Perception, Decision-making and Coping with Genetic Diseases

Genetic counseling is psychoeducational-based counseling. Decision-making in genetic counseling straddles the tension between motivated reasoning and a wish for a favorable outcome. When genetic counseling occurs, clients are typically in acute distress. Within this context, complex genetic knowledge, probability principles and recurrence risks, variability of gene expression, and various prophylactic interventions must be clearly conveyed by the genetic counselor to his or her client. Thus, it is not surprising that researchers have demonstrated that genetic decision-making is complicated by the client's inability to understand principles of probability, motivation to have more children (Shiloh & Saxe, 1989), parity and family history (Ekwo, Kim, & Gosselink, 1987) and counselor presentation styles of objective numeric risk information (Kessler & Levine, 1987; Marteau, 1989; Marteau, Plenicar, & Kidd, 1993; McNeil, Pauker, Sox, & Tversky, 1982).

Decision-making associated with genetic information revolves around major life choices. To this end, most people have had no experience and may feel ill-equipped and/or unjustified to make these decisions. Additionally, the gravity of these decisions are embedded in profound moral and interpersonal dilemmas and impact life long decisions for the individual, family members, and future generations such as: child-bearing (having a child (or another child) considered at-risk for a genetic disease); marriage (e.g., consanguinity), and; infertility and/or decreasing genetic risks when both spouses care the same gene for a genetic disease (e.g, artificial insemination). Unfortunately, the distress associated with genetic counseling is in part due to the uncertain and probabilistic nature of genetic information coupled with a misguided presupposition that a decision begins a cascade of consecutive choices. The decision-making process is complex. For example, the genetic counselor and/or client may mistakenly assume that a decision to use prenatal diagnosis implies a choice to terminate the pregnancy of an affected fetus when in fact these are separate decisions. Realistically, a client must decide whether: (1) she should undergo genetic prenatal tests that carry procedural risks; (2) continue or terminate a pregnancy after learning that the fetus is affected or not affected with a genetic disease, and; (3) if the fetus is carried to term, the planning and implementation of the necessary adjuvant therapies to meet the needs of the child and family.

Individual coping strategies are related to an person's lifestyle, experiences with

health problems, degree of self-blame, beliefs, and cultural backgrounds. Kessler (1984) has described cognitive coping strategies people use when confronted with issues involving genetic diseases as a means of restoring hope and regaining a sense of personal control. He has argued that people: (1) seek declarative knowledge about the specific genetic disease; (2) develop new coping strategies by making decisions among various choices of action; and (3) become better informed of available technology to minimize their uncertainty and distress (e.g., prenatal diagnosis, bone marrow transplant as a treatment for a disease).

Family dynamics can also influence coping strategies (Kronenberger & Thompson, 1992). Strauss (1988) showed that families holding a more accepting meaning of their child's genetic condition (God's will) displayed better adaptation than families accepting a negative (punishment) meaning. Conversely, Kessler and Bloch (1989) reported how, in the instance of adult onset genetic diseases, dysfunctional families can stigmatize individual family members through preselection - the identification of a asymptomatic family member predicted to become an individual affected with the genetic disease. The results can range from lowered self-esteem, isolation, or in extreme cases suicide.

Implications of Genetic Counseling for Psychology

More and more people are touched by genetic information daily. Even with the technological accomplishments generated by the Human Genome Initiative, genetic information will continue to produce uncertainty. It is this uncertainty that gives rise to many of the complex clinical dilemmas facing genetic and psychology professionals today. How does genetic information influence the development of personality and identity formation of people with genetic diseases compared to individuals at risk? How does knowing genetic information and the potential of having a life-threatening disease influence family relationships and life expectations? Genetic diseases challenge one's understanding who is self. Unlike other diseases that are experienced as ego-alien, genetic conditions are experienced as a constitutional part of one's self. No option exists to diminish this threat by projecting it onto an external cause.

Thus, geneticists and psychologists must move beyond the Cartesian separation of the mind (psychology) and body (genetics) embodied separately in each profession. An integrated mind-body orientation must be adopted which more realistically contextualizes a professional's experiences in hope of better understanding who they are and the people they treat. This approach will allow geneticists to move beyond their psychoeducational stance and view clients more than biological DNA units. This perspective will also free psychologists to go beyond the intrapsychic, existential meanings of identity and integrate a genetic understanding of self as part of the gestalt of identity formation.

Overall, psychologists will need to better attend to the clinical presentation of genetic issues with clients (e.g., individuals, couples and family members who have complex health histories and be at risk for genetic diseases). Given the logistics and training limitations of genetic counselors, psychologists must be prepared to work with individuals, couples and families to explore how the impact of genetic

information influences coping, adjustment, and understanding of oneself. They must be open to exploring the intrapsychic issues related to uncertainty, risk perception and decision-making. Genetics and psychology have met at a crossroad. Is the road to be traveled?

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